Questions EAnswers about ...

Autoimmunity

National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) National Institutes of Health Public Health Service • U.S. Department of Health and Human Services

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Autoimmunity

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This booklet answers questions about autoimmunity. To learn more about autoimmunity, your immune system, or autoimmune diseases, speak with your doctor. You can also check out the Appendices on pages 22–29.

What Is Autoimmunity?

When your body is attacked—perhaps by a virus or germs on a nail you stepped on—your immune system defends you. It sees and kills the germs that might hurt you.

But when the system doesn't work right, this process can cause harm. Immune cells can mistake your body's own cells as invaders and attack them. This "friendly fire" can affect almost any part of the body. It can sometimes affect many parts of the body at once. This is called autoimmunity (meaning self-immunity).

What Causes Autoimmunity?

No one knows why the immune system treats some body parts like germs. We do know that you can't catch autoimmune diseases from another person.

Most scientists think that our genes and things in the environment are involved. If you have a certain gene or combination of genes, you may be at higher risk for autoimmune disease. But you won't get the disease until something around you turns on your immune system. This may

include the sun, infections, drugs, or, in some women, pregnancy.

What Kinds of Problems Are Caused by Autoimmunity?

Autoimmunity can affect almost any organ or body system. The exact problems one has with autoimmunity (or its diseases) depends on which tissues are targeted.

If the skin is the target, you may have skin rashes, blisters, or color changes. If it's the thyroid gland, you may be tired, gain weight, be more sensitive to cold, and have muscle aches. If it's the joints, you may have joint pain, stiffness, and loss of function.

You may know which organ or system is affected from the start. But you may not know the site of the attack. In many people, the first symptoms are fatigue, muscle aches, and low fever.

Where Does Autoimmunity Strike?

Because autoimmune diseases can affect almost any organ or system of the body, one way to group them is by the body system(s) they attack. The following is a list (not inclusive) of body systems and the autoimmune diseases that can affect them.

Blood and blood vessels

- · Autoimmune hemolytic anemia
- · Pernicious anemia
- · Polyarteritis nodosa
- Systemic lupus erythematosus
- Wegener's granulomatosis

Digestive tract (including the mouth)

- Autoimmune hepatitis
- · Behçet's disease
- Crohn's disease
- · Primary bilary cirrhosis
- Scleroderma
- Ulcerative colitis

Eyes

- · Sjögren's syndrome
- Type 1 diabetes mellitus
- Uveitis

Glands

- · Graves' disease
- Thyroiditis
- Type 1 diabetes mellitus

Heart

- Myocarditis
- Rheumatic fever
- Scleroderma
- Systemic lupus erythematosus

Joints

- · Ankylosing spondylitis
- · Rheumatoid arthritis
- · Systemic lupus erythematosus

Kidneys

- · Glomerulonephritis
- Systemic lupus erythematosus
- Type 1 diabetes mellitus

Lungs

- · Rheumatoid arthritis
- Sarcoidosis
- Scleroderma
- · Systemic lupus erythematosus

Muscles

- Dermatomyositis
- Myasthenia gravis
- Polymyositis

Nerves and brain

- · Guillain-Barré syndrome
- Multiple sclerosis
- · Systemic lupus erythematosus

Skin

- Alopecia areata
- Pemphigus/pemphigoid
- Psoriasis
- Scleroderma
- Systemic lupus erythematosus
- Vitiligo

How Are Autoimmune Diseases Diagnosed?

Autoimmune diseases often don't show a clear pattern of symptoms at first. So diagnosing them can be hard. But with time, a diagnosis can usually be made by using:

- Medical history—The doctor will ask about your symptoms and how long you have had them. Your symptoms may not point to one disease. But they can be a starting point for your doctor. You should tell your doctor if you have a family member with autoimmune disease. You may not have the same disease as your family member. But having a family history of any autoimmune disease makes you more likely to have one.
- Physical exam—During the exam, the doctor will check for any signs. Inflamed joints, swollen lymph nodes, or discolored skin might give clues.
- Medical tests—No one test will show that you have an autoimmune disease. But doctors may find clues in a blood sample. For example, people with lupus or rheumatoid arthritis often have certain autoantibodies in their blood. Autoantibodies are blood proteins formed against the body's own parts.

Not all people with these diseases have these autoantibodies. And some people without autoimmune disease do have them. So blood tests alone may not always help. But if a person has disease symptoms and autoantibodies, the doctor can be more sure of a diagnosis.

The key is patience. Your doctor may be able to diagnose your condition quickly based on your history, exam, and test results. But the process often takes time. It may take several visits to find out exactly what's wrong and the best way to treat it.

How Are Autoimmune Diseases Treated?

Autoimmunity takes many forms. There are also many treatments for it. Treatment depends on the type of disease, how severe it is, and its symptoms. Generally, treatments have one of three goals:

- Relieving symptoms—If your symptoms bother you, your doctor may suggest treatments that give some relief. Relieving symptoms may be as simple as taking a drug for pain relief. It may also be as involved as having surgery.
- Preserving organ function—When autoimmune diseases threaten organs, treatment may be needed to prevent damage. Such treatments may include drugs to control an inflamed kidney in people with lupus. Insulin injections can regulate blood sugar in people with diabetes. These treatments don't stop the disease. But they can save organ function. They can also help people live with disease complications.

 Targeting disease mechanisms—Some drugs may also be used to target how the disease works. In other words, they can suppress the immune system. These drugs include cyclophosphamide (Cytoxan*) and cyclosporine (Neoral and Sandimmune). The same immune-suppressing drug may be used for many diseases.

Your doctor may not prescribe a treatment. If your symptoms are mild, the risks of treatment may be worse than the symptoms. You may choose to put off treatment for now. But you should watch for signs that your disease is progressing. Visit your doctor regularly. You need to catch changes before they lead to serious damage.

What Types of Doctors Treat Autoimmune Diseases?

Treatments for autoimmune diseases vary. So do the types of doctors who provide them.

For some people, one doctor will be enough to manage their disease. Others may require a team approach. One doctor might coordinate and give care, and others would treat specific organ problems. For example, a person with lupus might be seen by a rheumatologist. But that person might

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also see a nephrologist for related kidney problems and a dermatologist for skin problems.

Specialists you may need to see include:

- A rheumatologist, who treats arthritis and other rheumatic diseases. These include scleroderma and systemic lupus erythematosus (lupus or SLE).
- An endocrinologist, who treats gland and hormone problems. These include diabetes and thyroid disease.
- A neurologist, who treats nerve problems. These include multiple sclerosis and myasthenia gravis.
- A hematologist, who treats diseases that affect the blood. These include pernicious anemia and autoimmune hemolytic anemia.
- A gastroenterologist, who treats problems with the digestive system. These include Crohn's disease and ulcerative colitis.
- A dermatologist, who treats problems of the skin, hair, and nails. These include psoriasis, pemphigus/ pemphigoid, and alopecia areata.
- A nephrologist, who treats kidney problems. These include glomerulonephritis, inflamed kidneys associated with lupus.

What Are Some Other Problems Related to Autoimmune Diseases?

Having a chronic disease can affect almost every part of your life. The problems you might have with an autoimmune disease vary. They may include:

- How you look and your self-esteem—Depending on your disease, you may have discolored or damaged skin or hair loss. Your joints may look different.

 These can all affect how you look and your self-esteem. Such problems can't always be prevented.

 But their effects can be reduced with treatment.

 Cosmetics, for example, can hide a skin rash.

 Surgery can correct a malformed joint.
- Caring for yourself—Painful joints or weak muscles
 can make it hard to do simple tasks. You may have
 trouble climbing stairs, making your bed, or brushing
 your hair. If doing daily tasks is hard, talk with a
 physical therapist. The therapist can teach you exercises to improve strength and function. An occupational therapist can show you new ways to do things
 or tools to make tasks easier. Sometimes regular
 exercise or simple devices can help you do more
 things on your own.
- Family relationships—Family members may not understand why you don't have energy to do things you used to do. They may even think you are just being lazy. But they may also be overly concerned

and eager to help you. They may not let you do the things you can do. They may even give up their own interests to be with you. Learn as much as you can about your disease. Share what you learn with your family. Involve them in counseling or a support group. It may help them better understand the disease and how they can help.

- Sexual relations—Sexual relationships can also be affected. For men, diseases that affect blood vessels can lead to problems with erection. In women, damage to glands that produce moisture can lead to vaginal dryness. This makes intercourse painful. In both men and women, pain, weakness, or stiff joints may make it hard for them to move the way they once did. They may not be sure about how they look. Or they may be afraid that their partner will no longer find them attractive. With communication, good medical care, and perhaps counseling, many of these issues can be overcome or at least worked around.
- Pregnancy and childbearing—In the past, women with some autoimmune diseases were told not to have children. But better treatments and understanding have changed that advice. Autoimmune diseases can affect pregnancy, and pregnancy can affect autoimmune diseases. But women with many such diseases can safely have children. How a pregnancy turns out can vary by disease and disease severity. If you have an autoimmune disease, you should consult your doctor about having children.

What Research Is Being Conducted To Help People With Autoimmune Diseases?

The National Institutes of Health (NIH) supports research in autoimmune diseases. Here are a few examples:

- Rheumatoid arthritis—The National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) and the North American Rheumatoid Arthritis Consortium will study 1,000 siblings with rheumatoid arthritis. Scientists will look at gene material to find and identify parts of DNA involved in the disease. They will test for proteins called rheumatoid factor in the blood. And they will look at x rays of the joints. This work will provide basic facts about the genetics of the disease.
- Systemic lupus erythematosus (SLE)—NIAMSsupported scientists are studying whether women with lupus can safely take oral contraceptives or hormone replacement therapy. Previous research suggests that female hormones may contribute to the disease or make it worse.
- Lupus nephritis—One NIAMS project is testing a drug that may be less toxic than the drugs now used for lupus nephritis (kidney disease caused by lupus).
- Vitiligo—With NIAMS support, scientists are studying genes from pairs of siblings affected by this skin

pigmentation disorder. They hope to find genes that may cause vitiligo and learn how they affect the skin.

- Type 1 diabetes—Researchers supported by the National Institute of Diabetes and Digestive and Kidney Diseases have found a way to identify people who are likely to get type 1 diabetes (formerly known as juvenile diabetes). They are now testing ways to prevent these people from getting the disease.
- Multiple sclerosis—Scientists supported by the National Institute of Neurological Disorders and Stroke are looking at the autoimmune system, infectious agents, and genes as culprits in multiple sclerosis (MS). Such studies strengthen the theory that MS comes from a number of factors rather than a single one. Studies use magnetic resonance imaging to see how MS lesions evolve in the brain's white matter. Research has shown that MS has no bad effects on pregnancy, labor, or delivery. In fact, the stabilizing or remission of symptoms during pregnancy may be due to changes in a woman's immune system that allow her to carry a baby.
- Multiple autoimmune diseases—The National
 Institute of Allergy and Infectious Diseases is supporting clinical trials of drugs that prevent the
 immune system from attacking healthy cells. The
 Institute wants to see if they are safe and useful.
 Such drugs may prove helpful for treating a
 number of autoimmune diseases.

Where Can People Find More Information About Autoimmunity?

Various parts of the NIH provide information on different aspects of autoimmune diseases. Many nonprofit organizations have patient resources, local chapters, and support groups. Your own doctor can best tell you about your own medical condition.

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www.niaid.nih.gov/publications

and

www.niaid.nih.gov/clintrials/default.htm (for clinical

trials information)

National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)

National Diabetes Information Clearinghouse (NDIC)

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National Institute of Neurological Disorders and Stroke

Office of Scientific and Health Reports

P.O. Box 5801

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www.ninds.nih.gov

NIH Clinical Center

Patient Recruitment and Public Liaison—for specific

NIH clinical trials information

Building 60

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Phone: 800-411-1222 (free of charge)

TTY: 866-411-1010

E-mail: prpl@mail.cc.nih.gov

www.clinicalstudies.info.nih.gov/referring patient.html

Office of Rare Diseases

Building 31, Room 1B19

31 Center Drive

Bethesda, MD 20892

Phone: 301-402-4336

www.rarediseases.info.nih.gov/ord

Other Resources Sponsored by the U.S. Department of Health and Human Services

Combined Health Information Database http://chid.nih.gov

National Health Information Center

Phone: 301-565-4167 or 800-336-4797 (free of charge)

Health Finder: www.healthfinder.gov

Other Organizations

American Academy of Dermatology

930 N. Meacham Road

P.O. Box 4014

Schaumburg, IL 60168-4014

Phone: 847-330-0230 or

888-462-DERM (3376) (free of charge)

Fax: 847-330-0050

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American Academy of Orthopaedic Surgeons

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American College of Rheumatology

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American Autoimmune Related Diseases Association

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East Detroit, MI 48201

Literature requests: 800-598-4668 (free of charge)

Phone: 586-776-3900

Fax: 586-776-3903

E-mail: aarda@aol.com

www.aarda.org

American Behçet's Disease Association

P.O. Box 15247

Chattanooga, TN 37415-0240

Phone: 800-7-BEHCET (723-4238) (free of charge)

www.behcets.com

American Diabetes Association

Attn: Customer Service

1701 N. Beauregard Street

Alexandria, VA 22311

Phone: 800-DIABETES (342-2383) (free of charge)

www.diabetes.org

American Liver Foundation

75 Maiden Lane, Suite 603

New York, NY 10038

Phone: 973-256-2550 or 800-443-7222 (free of charge)

www.liverfoundation.org

Arthritis Foundation

1330 West Peachtree Street

Atlanta, GA 30309

Phone: 800-283-7800 (free of charge)

www.arthritis.org

Crohn's and Colitis Foundation of America

National Headquarters

386 Park Avenue South, 17th Floor

New York, NY 10016-8804

Phone: 800-932-2423 (free of charge)

800-343-3637 (Warehouse)

www.ccfa.org

■ Guillain-Barré Syndrome Foundation International

P.O. Box 262

Wynnewood, PA 19096

Phone: 610-667-0131

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E-mail: gbint@ix.netcom.com

www.guillain-barre.com

Juvenile Diabetes Research Foundation International

120 Wall Street

New York, NY 10005-4001

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800-JDF-CURE (533-2873) (free of charge)

Fax: 212-785-9595

E-mail: info@jdrf.org

www.jdf.org

Lupus Foundation of America

1300 Piccard Drive, Suite 200

Rockville, MD 20850-4303

Phone: 301-670-9292 or 800-558-0121 (free of charge)

www.lupus.org

Myasthenia Gravis Foundation of America

5841 Cedar Lake Road, Suite 204

Minneapolis, MN 55416

Phone: 952–545–9438 or 800–541–5454 (free of charge)

Fax: 952-545-6073

E-mail: myasthenia@myasthenia.org

www.myasthenia.org

Myositis Association of America

755 Cantrell Avenue, Suite C

Harrisonburg, VA 22801

Phone: 540-433-7686

Fax: 540-432-0206

E-mail: maa@myositis.org

www.myositis.org

National Adrenal Diseases Foundation

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E-mail: nadfmail@aol.com

www.medhelp.org/nadf

National Alopecia Areata Foundation

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National Multiple Sclerosis Society

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National Organization for Rare Diseases

P.O. Box 8923

New Fairfield, CT 06812-1783

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Fax: 203-746-6481

www.rarediseases.org

National Pemphigus Foundation

828 San Pablo Avenue, Suite 203

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Phone: 510-527-4970

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E-mail: pvnews@aol.com

www.pemphigus.org

National Psoriasis Foundation

6600 SW 92nd Avenue, Suite 300

Portland, OR 97223-7195

Phone: 503-244-7404 or 800-723-9166 (free of charge)

Fax: 503-245-0626

E-mail: getinfo@npfusa.org

www.psoriasis.org

National Vitiligo Foundation

611 South Fleishel Avenue

Tyler, TX 75701

Phone: 903-531-0074

Fax: 903-525-1234

E-mail: vitiligo@trimofran.org

www.nvfi.org

Scleroderma Foundation

12 Kent Way, Suite 101

Bayfield, MA 01922

Phone: 978-463-5843

Info line: 800-722-HOPE (4673) (free of charge)

Fax: 978-463-5809

E-mail: sfinfo@scleroderma.org

www.scleroderma.org

Scleroderma Research Foundation

2320 Bath Street, Suite 315

Santa Barbara, CA 93105

Phone: 805-563-9133 or

800-441-CURE (2873) (free of charge)

Fax: 805-563-2402

E-mail: srfcure@srfcure.org

www.srfcure.org

Sjögren's Syndrome Foundation

8120 Woodmont Avenue, Suite 530

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800-4-SJOGRENS (475-6473) (free of charge)

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E-mail: ssf@sjogrens.org

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Spondylitis Association of America

P.O. Box 5872

Sherman Oaks, CA 91413

Phone: 800-777-8189 or 888-777-1594 (free of charge)

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SLE Foundation, Inc.

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■ Thyroid Foundation of America, Inc.

410 Stuart Street Boston, MA 02116

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Appendix I: Glossary of Terms

Antibodies—Special proteins produced by the body's immune system that help fight and destroy viruses, bacteria, and other foreign substances that invade the body.

Antigen—A substance (usually foreign) that stimulates the immune response. In people with autoimmune disease, the body's own cells may be seen as antigens.

Autoantibodies—Abnormal antibodies that attack parts of the body, causing autoimmune disease.

Autoimmune disease—A disease that occurs when the immune system turns against parts of the body it is designed to protect.

Fever—A rise in body temperature caused by the immune system's response to infection or disease.

Immune response—The reaction of the immune system against foreign substances. When the reaction occurs against the body's own cells or tissues, it is called an auto-immune reaction.

Immune system—A complex system that normally protects the body from infections. The immune system consists of a group of cells, the chemicals that control those cells, and the chemicals that those cells release.

Immunosuppressive drugs—Drugs that suppress the immune response and can be used to treat autoimmune disease. Unfortunately, because normal immunity is also suppressed with these drugs, they leave the body at risk for infection.

Infection—Invasion of the body tissues by bacteria or other tiny organisms that cause illness.

Inflammation—A reaction of tissues to injury or disease, typically marked by four signs: swelling, redness, heat, and pain.

Trigger—Something that either sets off a disease in people who are genetically predisposed to developing the disease, or that causes a certain symptom to occur in a person who has a disease. For example, sunlight can trigger rashes in people with lupus.

Appendix II: Glossary of Diseases

Autoimmunity plays a role in more than 80 diseases. Following are brief descriptions of some of the many diseases in which autoimmunity may be involved.

Note: Because the specific causes of many diseases are unknown, there is debate among scientists about whether some of these are truly autoimmune diseases. Your own doctor may classify some of these diseases differently.

Alopecia areata—A disorder in which the immune system attacks the hair follicles, causing loss of hair on the scalp, face, and other parts of the body.

Ankylosing spondylitis—A rheumatic disease that causes inflamed joints in the spine and sacroiliac (the joints that connect the spine and the pelvis) and, in some people, inflamed eyes and heart valves.

Arthritis—A general term for more than 100 different diseases that affect the joints. Many forms of arthritis and related conditions are believed to have an autoimmune component.

Autoimmune hemolytic anemia—A condition in which immune system proteins attack the red blood cells, resulting in fewer of these oxygen-transporting cells.

Autoimmune hepatitis—A disease in which the body's immune system attacks liver cells, causing inflammation. If not stopped, inflammation can lead to cirrhosis (scarring and hardening) of the liver and eventually liver failure.

Behçet's disease—A condition characterized by sores in the mouth and on the genitals and by inflammation in parts of the eye. In some people, the disease also results in inflammation of the joints, digestive tract, brain, and spinal cord.

Crohn's disease—An inflammatory disease of the small intestine or colon that causes diarrhea, cramps, and excessive weight loss.

Dermatomyositis—A rare autoimmune disease that causes patchy red rashes around the knuckles, eyes, and other parts of the body along with chronic inflammation of the muscles. It may occur along with other autoimmune diseases such as rheumatoid arthritis or systemic lupus erythematosus.

Diabetes mellitus, type 1—A condition in which the immune system destroys insulin-producing cells of the pancreas, making it impossible for the body to use glucose (blood sugar) for energy. Type 1 diabetes usually occurs in children and young adults.

Glomerulonephritis—Inflammation of the kidney's tiny filtering units, which in severe cases can lead to kidney failure. Graves' disease—An autoimmune disease of the thyroid gland that results in the overproduction of thyroid hormone. This causes such symptoms as nervousness, heat intolerance, heart palpitations, and unexplained weight loss.

Guillain-Barré syndrome—A disorder in which the body's immune system attacks part of the nervous system, leading to numb, weak limbs and, in severe cases, paralysis.

Inflammatory bowel disease—The general name for diseases that cause inflammation in the intestine, the most common of which are ulcerative colitis and Crohn's disease.

Lupus nephritis—Damaging inflammation of the kidneys that can occur in people with lupus. If not controlled, it may lead to total kidney failure.

Multiple sclerosis—A disease in which the immune system attacks the protective coating called myelin around the nerves. The damage affects the brain and/or spinal cord and interferes with the nerve pathways, causing muscular weakness, loss of coordination, and visual and speech problems.

Myasthenia gravis—A disease in which the immune system attacks the nerves and muscles in the neck, causing weakness and problems with seeing, chewing, and/or talking.

Myocarditis—Inflamed and degenerating muscle tissue of the heart that can cause chest pain and shortness of breath. This can lead to congestive heart failure. **Pemphigus/pemphigoid**—An autoimmune disease of the skin characterized by itching and blisters.

Pernicious anemia—A deficiency of the oxygen-carrying red blood cells that often occurs in people with autoimmune diseases of the thyroid gland.

Polyarteritis nodosa—An autoimmune disease that causes inflammation of the small and medium-sized arteries. This leads to problems in the muscles, joints, intestines, nerves, kidney, and skin.

Polymyositis—A rare autoimmune disease characterized by inflamed and tender muscles throughout the body, particularly those of the shoulder and hip girdles.

Primary biliary cirrhosis—A disease that slowly destroys the bile ducts in the liver. When the ducts are damaged, bile (a substance that helps digest fat) builds up in the liver and damages liver tissue.

Psoriasis—A chronic skin disease that occurs when cells in the outer layer of the skin reproduce faster than normal and pile up on the skin's surface. This results in scaling and inflammation. An estimated 10 to 30 percent of people with psoriasis develop an associated arthritis called psoriatic arthritis.

Rheumatic fever—A disease that can occur following untreated streptococcus (strep) infection. It most often affects children, causing painful, inflamed joints and, in some cases, permanent damage to heart valves.

Rheumatoid arthritis—A disease in which the immune system is believed to attack the linings of the joints. This results in joint pain, stiffness, swelling, and destruction.

Sarcoidosis—A disease characterized by granulomas (small growths of blood vessels, cells, and connective tissue) that can lead to problems in the skin, lungs, eyes, joints, and muscles.

Scleroderma—An autoimmune disease characterized by abnormal growth of connective tissue in the skin and blood vessels. In more severe forms, connective tissue can build up in the kidneys, lungs, heart, and gastrointestinal tract, leading in some cases to organ failure.

Sjögren's syndrome—A condition in which the immune system targets the body's moisture-producing glands, leading to dryness of the eyes, mouth, and other body tissues.

Systemic lupus erythematosus—An autoimmune disease, primarily of young women, that can affect many parts of the body, including the joints, skin, kidneys, heart, lungs, blood vessels, and brain.

Thyroiditis—An inflammation of the thyroid gland that causes the gland to become underactive. This results in symptoms such as fatigue, weakness, weight gain, cold intolerance, and muscle aches.

Ulcerative colitis—A disease that causes ulcers in the top layers of the lining of the large intestine. This leads to abdominal pain and diarrhea.

Uveitis—The inflammation of structures of the inner eye, including the iris (the colored tissue that holds the lens of the eye) and the choroid plexus (a network of blood vessels around the eyeball). Uveitis occurs with some rheumatic diseases, including ankylosing spondylitis and juvenile rheumatoid arthritis.

Vitiligo—A disorder in which the immune system destroys pigment-making cells called melanocytes. This results in white patches of skin on different parts of the body.

Wegener's granulomatosis—An autoimmune disease that damages the small and medium-sized blood vessels throughout the body, resulting in disease in the lungs, upper respiratory tract, and kidneys.

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